

# Mystery still unresolved: Untouched “Blue heart” presenting at 40yrs of age

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## ABSTRACT

The modern-day surgical techniques and strategies have changed the outlook of patients with dextro-transposition of great arteries (d-TGA). The survival of an unrepaired d-TGA into late adulthood is difficult to explain. Even when large intracardiac shunts are present, it still remains a lethal cyanotic congenital heart disease if it is not surgically corrected soon after birth. Here, we report an extremely rare case of d-TGA presenting at 40 years of age, with moderately elevated pulmonary artery pressures and relatively stable symptoms.

**Keywords:** Adult congenital heart disease, cyanotic heart disease, transposition of great arteries

## INTRODUCTION

It is extremely rare for an unrepaired dextro-transposition of great arteries (d-TGA) to present in adulthood, even in the presence of large intracardiac shunts. Here, we present such a rare case presenting at 40 years of age, with relatively stable symptoms.

## CASE REPORT

A 40-year-old female, presented to our outpatient department, with complaints of dyspnea for 6 months (NYHA II). She denied any other cardiac symptoms and was not on any cardiac medications. She was not known to have any heart disease with apparently normal childhood history. There was no history of rheumatic fever or any significant cardiac history in the family. She had two uncomplicated pregnancies at the age of 25 and 32 years. She was found to have uniform central cyanosis, clubbing Grade II with saturations of 87% (preductal), and 88% (postductal), respectively. The jugular venous pulse showed prominent a wave. Blood pressure was 145/90 mmHg in the right arm. There was mild cardiomegaly, with right ventricular type of

apex. The first sound was normal with wide and fixed split-second sound. The pulmonary component was louder than aortic component. There was no additional S3/S4. There was a grade 2/6 ejection systolic murmur at pulmonary outflow.

The chest radiograph [Figure 1] showed enlarged cardiac silhouette, dilated main pulmonary artery (PA) with pulmonary plethora. A 12-lead Electrocardiogram [Figure 2] showed clockwise loop, sinus rhythm, right axis deviation of QRS vector with enlargement of the right atrium and ventricle.

Two-dimensional echocardiography images [Figure 3] showed large secundum atrial septal defect (ASD) with bidirectional flow. There was a small upper muscular ventricular septal defect (VSD) as well. The right ventricle (RV) was giving origin to aorta, and the left ventricle (LV) was giving origin to the PA [Figure 2]. The left ventricular thickness (posterior wall) measured 4.2 mm, suggesting regression. Tricuspid annular plane systolic excursion (TAPSE) was 22 mm. Although catheter study is not routinely indicated in d-TGAs, we proceeded

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with catheter study in view of the age of the patient to assess the operability and confirmation of the diagnosis. The oximetry [Table 1] showed almost equal saturations in the right and left ventricles with similar values in aorta and PA. Anterior ventriculogram [Figure 4] showed morphological RV giving origin to aorta, whereas posterior ventriculogram [Figure 4] showed morphological left ventricle giving origin to PA with confluent branches. There was a small upper muscular ventricular septal defect noted. Pulmonary angiogram [Figure 4] showed confluent and good-sized branch pulmonary arteries with good distal arborization (panel 5 and 6). LV and RV angiogram was suggestive of normal biventricular systolic function.

## DISCUSSION

The final diagnosis made was d-TGA, small upper muscular VSD, nonrestrictive ASD, and moderately elevated PA pressures. TGA is the most common form of cyanotic congenital heart disease presenting in the newborn period and comprises approximately 5% of all congenital heart defects.<sup>[1]</sup> The longest life expectancy of unoperated TGA that we saw in literature was in a 31-year-old Nigerian woman complicated by

Eisenmenger syndrome.<sup>[2]</sup> Unrepaired d-TGA surviving till 40 years of age is unusual, and we could not find any reported case on extensive literature review. Untreated, d-TGA is a fatal disease. From a large database between 1957 and 1964 in the state of California, a unique study performed by Liebman *et al.* has found that untreated, 29% of newborns with TGA died by the 1<sup>st</sup> week of life, 52% by the 1<sup>st</sup> month of life, and almost 90% were dead by the 1<sup>st</sup> year of life.<sup>[3]</sup> The natural history study by Shafer and Liebman *et al.* in the early 1960s found that no survival beyond 22 years of age.<sup>[3,4]</sup> The possible explanation for survival so far is the presence of an unrestrictive ASD which is providing adequate mixing, absence of significant pulmonary hypertension, and preserved RV systolic function. The absence of significant pulmonary hypertension in this patient can be explained by the absence of significant posttricuspid shunting.<sup>[5]</sup> The symptoms in this patient are secondary to the RV (systemic ventricle) diastolic dysfunction since

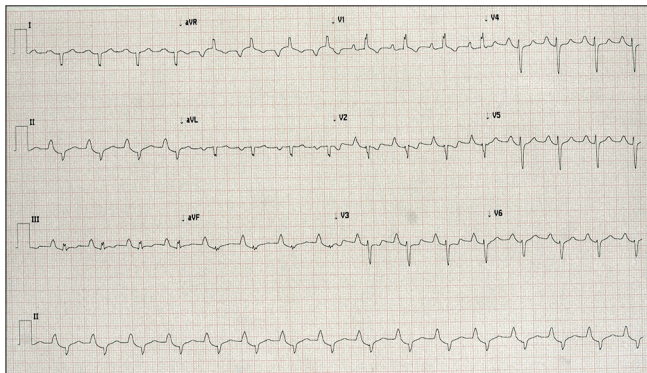


Figure 1: Electrocardiography shows sinus rhythm, clockwise loop, right axis deviation, right atrial, and right ventricular enlargement

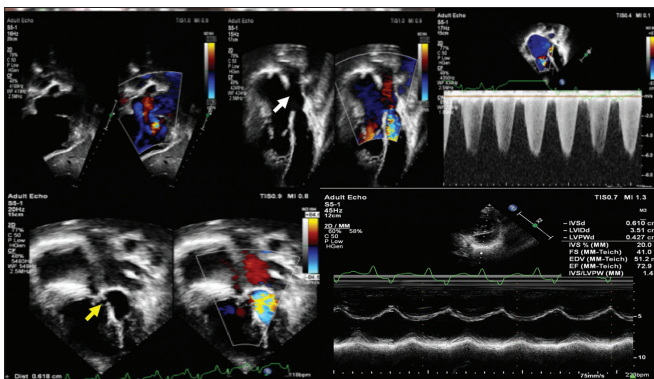


Figure 3: Two dimensional echocardiography: White arrows – large unrestrictive atrial septal defect, Yellow arrow – small restrictive ventricular septal defect, regressed left ventricle on short axis

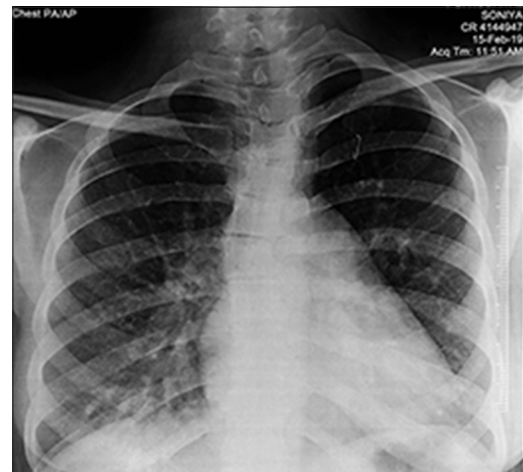


Figure 2: Chest X-ray shows cardiomegaly, dilated main pulmonary artery with pulmonary plethora and narrow superior mediastinum

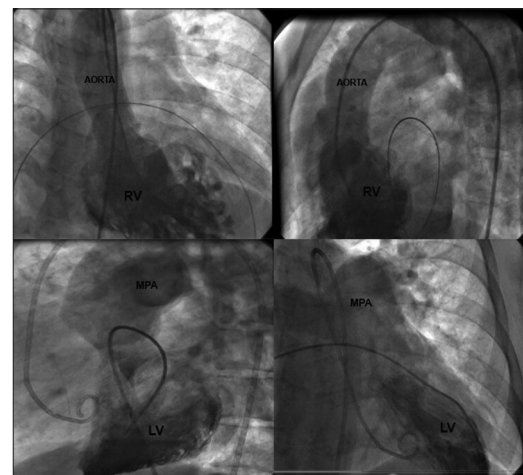


Figure 4: Left ventricle and right ventricle angiogram shows coarsely trabeculated ventricle (right ventricle) giving rise to aorta, and finally, trabeculated ventricle (left ventricle) was giving rise to pulmonary artery s/o transposition of great arteries

**Table 1: Hemodynamic data**

Pressures and Oximetry	Values	Mean pressures
Baseline pressure data		
Left atrial mean (mmHg)	12	
Right atrial mean (mmHg)	12	
RV systolic, EDP (mmHg)	150, 14	
Pulmonary artery (mmHg)	50/30	37
LV systolic, EDP (mmHg)	58, 10	
Ascending aorta (mmHg)	160/86	117
Descending thoracic aorta (mmHg)	168/84	112
Right femoral artery (mmHg)	170/80	100
Baseline oximetry		
Left innominate vein (%)	62	
Superior vena cava (%)	66	
Inferior vena cava (%)	70	
RV (%)	90	
Left pulmonary artery (%)	88	
Left upper PV (%)	100	
LV (%)	90	
Ascending aorta (%)	89	
Descending aorta (%)	89	
Right femoral artery (%)	88	

EDP: End-diastolic pressure, RV: Right ventricle, LV: Left ventricle, PV: Pulmonary vein

the patient had normal RV systolic function (TAPSE of 22 mm). The right ventricular diastolic dysfunction in this patient can be explained by elevated RV end-diastolic pressure (14 mmHg). Elevated serum brain natriuretic peptide (BNP) levels (626 pg/ml) suggest RV strain. The systemic hypertension has likely contributed to the diastolic dysfunction in this patient.<sup>[6]</sup> Since pulmonary pressures were only moderately elevated, shunts, SVR, and PVR were not calculated, and oxygen studies not performed. Regressed LV is suggested by D-shaped LV cavity, LV posterior wall thickness of <4.5 mm on echocardiography, and LV pressure/RV pressure of <0.85.<sup>[7]</sup> We have started this patient on diuretics and afterload reducing agents to be continued on follow-up.

In a study by Bajpai *et al.*, of six patients with d-TGA with a large posttricuspid shunt, for example, VSD, PDA, and aortopulmonary window, who had presented to the hospital late and were clinically judged to be worrisome for operability were administered oxygen to assess the operability, with an age range of 4 months–3 years (mean age: 1.1 years).<sup>[8]</sup> Of six patients studied, five patients appeared operable after the oxygen study.<sup>[8]</sup> However, after extensive literature review, we did not find any studies which address the natural history of d-TGA at this age as in our index case.

The low pressure in the LV will not be able to pump blood to serve the high pressure of systemic circulation if an arterial switch operation is performed in her. As she has lived till fourth decade without severe pulmonary hypertension and regressed LV, and we do not have great

literature support on long-term outcome after d-TGA surgery at the age of 40 years, we are not sure, offering surgical correction will alter the natural history.

#### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

#### Conflicts of interest

There are no conflicts of interest.

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